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Letter to the Editor

Major Central Nervous System Malformation in "Micromelic Dwarfism With Cone Epiphyses, Metaphyseal Dysplasia and Vertebral Segmentation Defects"

To the Editor:

Recently, we reported an apparently new type of spondylo-epiphyseal-metaphyseal dysplasia with "micromelic dwarfism, cone epiphyses, metaphyseal dysplasia and vertebral segmentation defects" in a 1.5-year-old male [Fryns et al., 1996].

On the occasion of a follow-up examination at the age of 2 years (weight 8.5 kg, height 65 cm, OFC 52.1 cm) we noted a persistent severe delay in motor development: the boy started to sit without support but marked hypotrophy of the lower legs was now evident. Neurological examination revealed spastic paraplegia with increased tone and increased reflexes with clonus and an exterior plantar response. Magnetic resonance imaging of the brain (Fig. 1a and b) showed major anomalies with corticosubcortical subdevelopment, hypoplasia of corpus callosum and enlarged ventricles. In addition, the sella turcica was small and the hypophyseal stalk elongated; the cervical spinal canal was narrow, most pronounced at the craniocervical junction, with kyphotic angulation of the cervical column.

While the latter anomalies are seen in several other short-limbed types of skeletal dysplasias, the corticosubcortical subdevelopment and the hypoplasia of the corpus callosum are primary developmental defects of the central nervous system as an apparently integral part of the syndrome.

REFERENCE

Fryns JP, Lorenzetti ME, Maroteaux P, Van den Berghe H (1996): Micromelic dwarfism with cone epiphyses, metaphyseal dysplasia, and vertebral segmentation defects. Am J Med Genet 61:164–167.

Jean-Pierre Fryns*
Center for Human Genetics
University Hospital Leuven
Leuven, Belgium

Fig. 1. **a**, **b**: Magnetic resonance imaging scan of the brain. Note marked corticosubcortical subdevelopment, hypoplasia of corpus callosum and enlarged ventricle.

* Correspondence to: Jean-Pierre Fryns, Center for Human Genetics, Herestraat 49, B-3000 Leuven, Belgium.

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